Journal: www.jogg.info
Originally Published: Volume 2, Issue 1 (Spring 2006)
Reference Number: 21.002

'SATIABLE CURIOSITY: NOW YOU SEE IT, NOW YOU DON'T: HETEROPLASMY IN MITOCHONDRIAL DNA

Author(s): Ann Turner

'SATIABLE CURIOSITY

Now You See It, Now You Don't: Heteroplasmy in Mitochondrial DNA

The 'Satiable Curiosity column is a regular feature in JoGG, suggesting projects where genetic genealogists and academic researchers can interact to their mutual benefit.

This issue of JoGG contains an article by David Pike, "Phylogenetic Networks for the Human mtDNA Haplogroup T," which proposes an intriguing and testable hypothesis about heteroplasmy. Might it explain the complex network diagrams formed by members of haplogroup T?[1]

Heteroplasmy – the coexistence of more than one mitochondrial DNA haplotype in one person – is a universal condition, although most of us are not even aware of its existence or potential genetic impact. Considering the thousands of mtDNA molecules inside each cell, each constantly being replenished, as well as the trillions of cells in the human body, it would be astonishing if every single mtDNA molecule were identical. Yet ordinary techniques for sequencing mtDNA do not detect the occasional mutation unless it reaches a level of approximately 20% of the total mitochondria. Thus, in essence, the signal of the minority is overwhelmed by the majority.

When cells divide, the daughter cells inherit a sampling of the mitochondria. As Figure 1 illustrates in a simplified way, the mixture of mtDNA in daughter cells will most often resemble the frequency found in the mother cell. However, it is also possible for the cell to have a higher or lower percentage of the mutant mtDNA.

Special techniques have been developed to quantify the amount of heteroplasmy at lower levels, down to 1% or even—with cloning techniques—down to the level of individual mitochondria.

Members of haplogroup T, who can recruit distant cousins descended from a common maternal ancestor, might provide good test cases for studying the level of heteroplasmy and how it persists or disappears over the course of several or many generations.

The results from such a study could provide useful insights not only for the study of population genetics, but also for the field of medicine.

Many mitochondrial diseases present a baffling picture of inheritance. For example, due to varying levels of heteroplasmy, one child may have full-blown symptoms of a disease while another child will have a lower percentage of heteroplasmic mtDNA and be disease-free.

Suggestions for future columns are welcome. Please submit them to Ann Turner, DNACousins@aol.com.

Ann Turner

[1] http://www.jogg.info/21/Pike.pdf

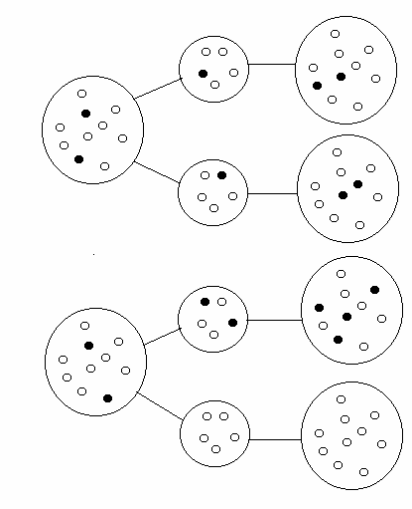

Figure 1 The percentage of mutant mtDNA may differ in daughter cells. In this illustration, 20% of the mitochondria in the mother cells (at the left) have mutant mtDNA (black circles), barely detectable by ordinary techniques. The mtDNA is divided among daughter cells in a random fashion, then the mitochondria multiply in the daughter cells to restore the original quantity. The percentage of mutant DNA in the daughter cells ranges from 0% to 40%.